

1/35

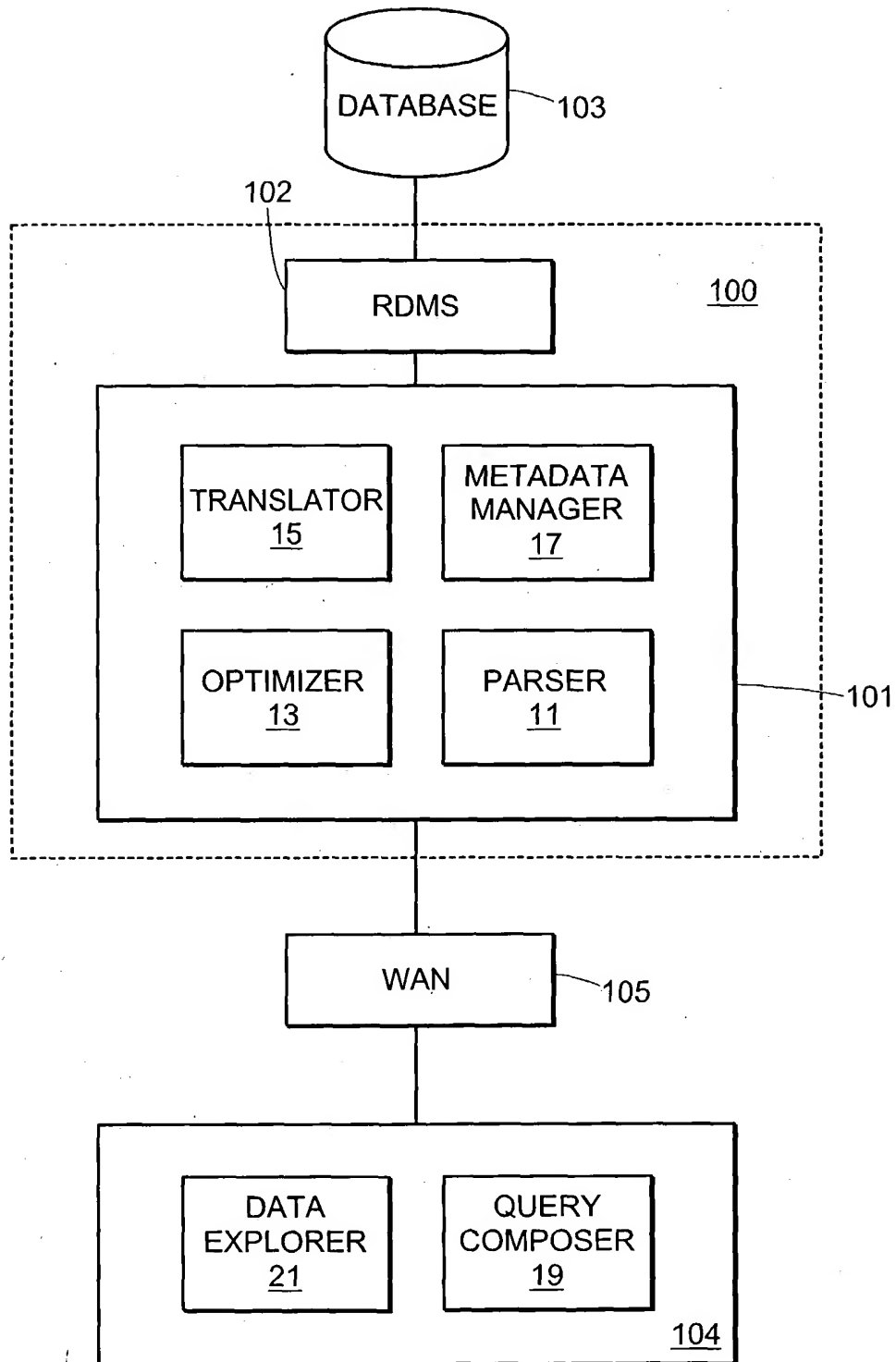


FIG. 1

2/35

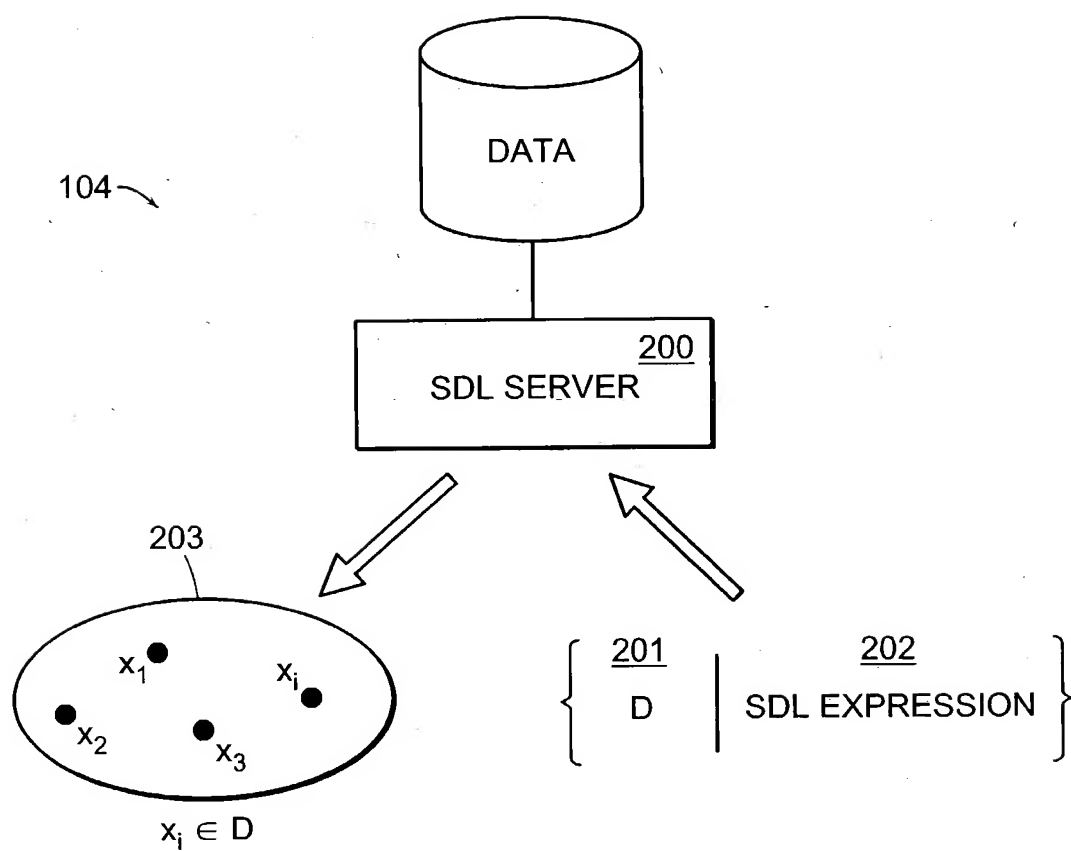


FIG. 2

3/35

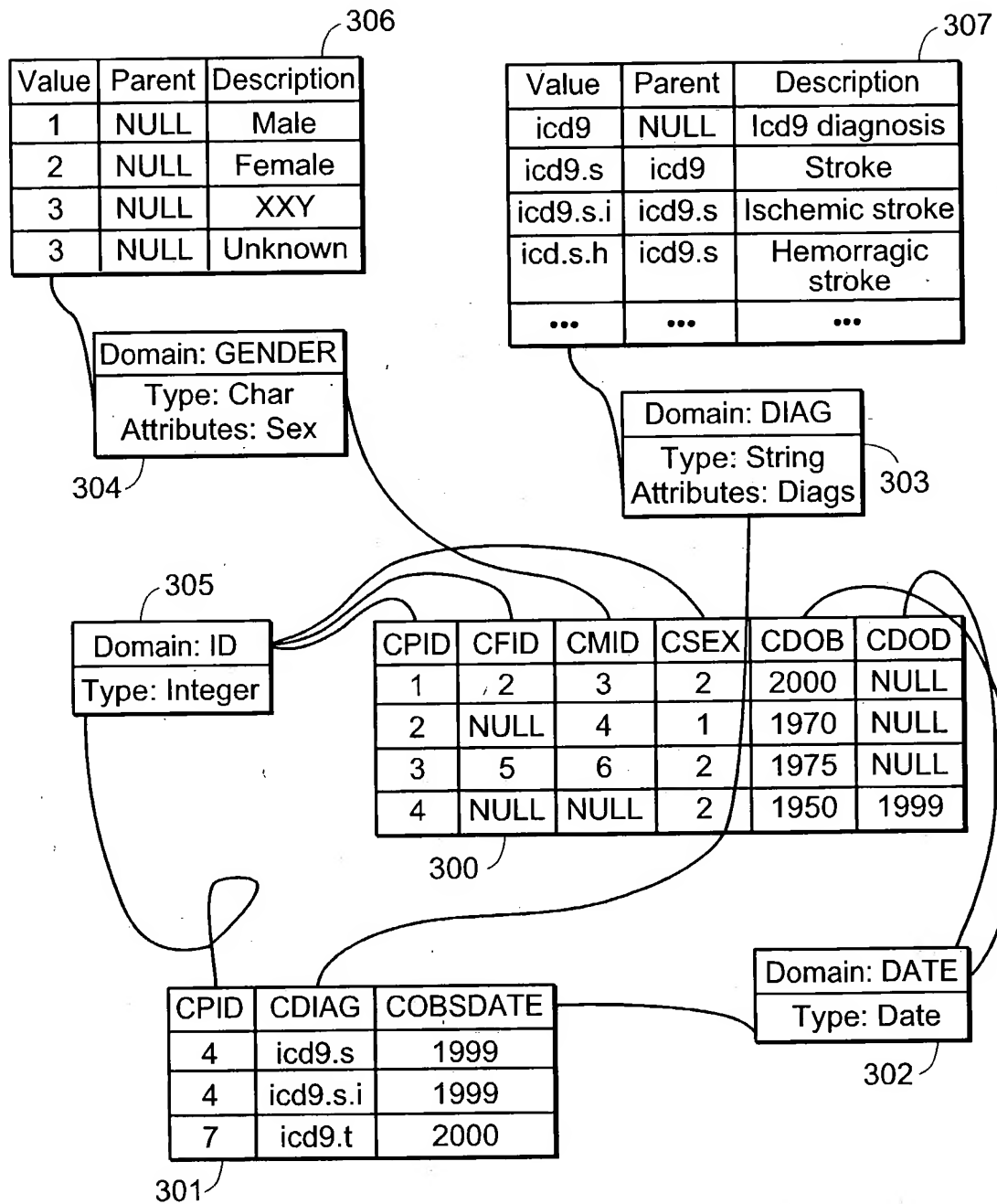


FIG. 3

4/35

400

	Domain	SQLtype	Hierarchy	Min	Max
303	DIAG	String	TDiags	NULL	NULL
304	GENDER	Char	TSex	NULL	NULL
302	DATE	Date	NULL	1900	2100
305	ID	Integer	NULL	0	NULL

401

Dimension	Domain
PID	ID
FID	ID
MID	ID
SEX	GENDER
DOB	DATE
DOD	DATE
OBSDATE	DATE
DIAG	DIAG

403

Dimension	Relation	Column_name	Multiplicity
PID	relation1	CPID	Unique
PID	relation2	CPID	Multiple
FID	relation1	CFID	Multiple
MID	relation1	CMID	Multiple
SEX	relation1	CSEX	Multiple
DOB	relation1	CDOB	Multiple
DOD	relation1	CDOD	Multiple
OBSDATE	relation2	COBSDATE	Multiple
DIAG	relation2	CDIAG	Multiple

402

Relation	SQLrelation	Inclusion_criteria
relation1	TableA	{PID, FID, MID, SEX, DOB, DOD}
relation2	TableB	Sex

FIG. 4

5/35

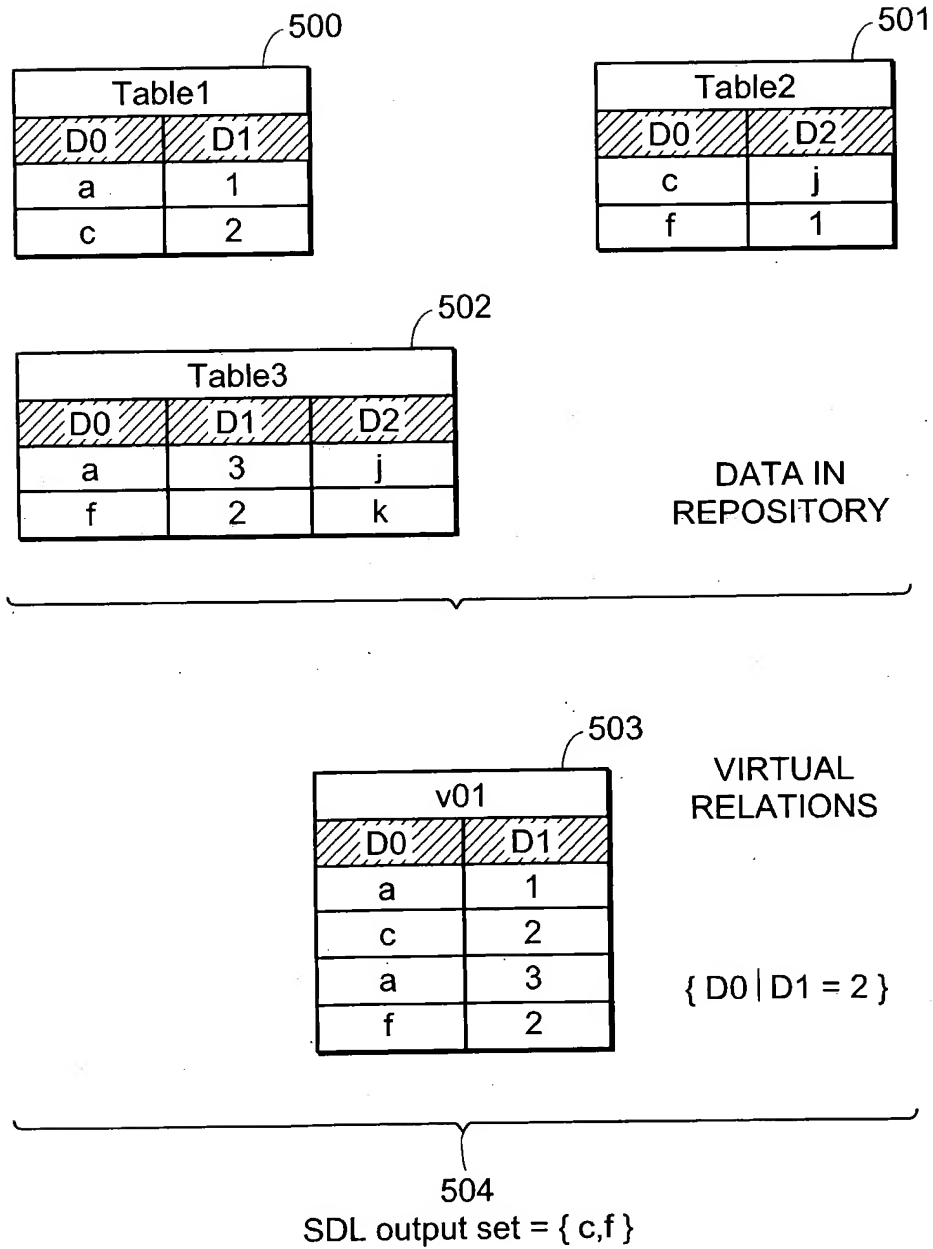


FIG. 5

6/35

600

PID	DIAG	HOSPID	DOCID	DATE
1	icd9.stroke	100	A	1966
2	icd9.x	200	B	2000

601

HOSPID	NAME	TYPE	REGION	ZIP
100	MGH	TH	Boston	02222
200	BWH	TH	Brookline	02115

602

PID	DATE	DIAG	HOSP. NAME	HOSP. TYPE	HOSP. REGION	HOSP. ZIP
1	1966	icd9.stroke	MGH	TH	Boston	02222
2	2000	icd9.x	BWH	TH	Brookline	02115

603

Relation	SQL relation	Inclusion_criteria
relation1	diags	{PID,DIAG,HOSPID,DOCID}
relation2	hospitals	{HOSPID,NAME,TYPE,REGION, ZIP}
relation3	vdiags	{PID,DIAG,HOSPID,DOCID}, {HOSP.NAME,HOSP.TYPE, HOSP.REGION,HOSP.ZIP}

604

FIG. 6

7/35

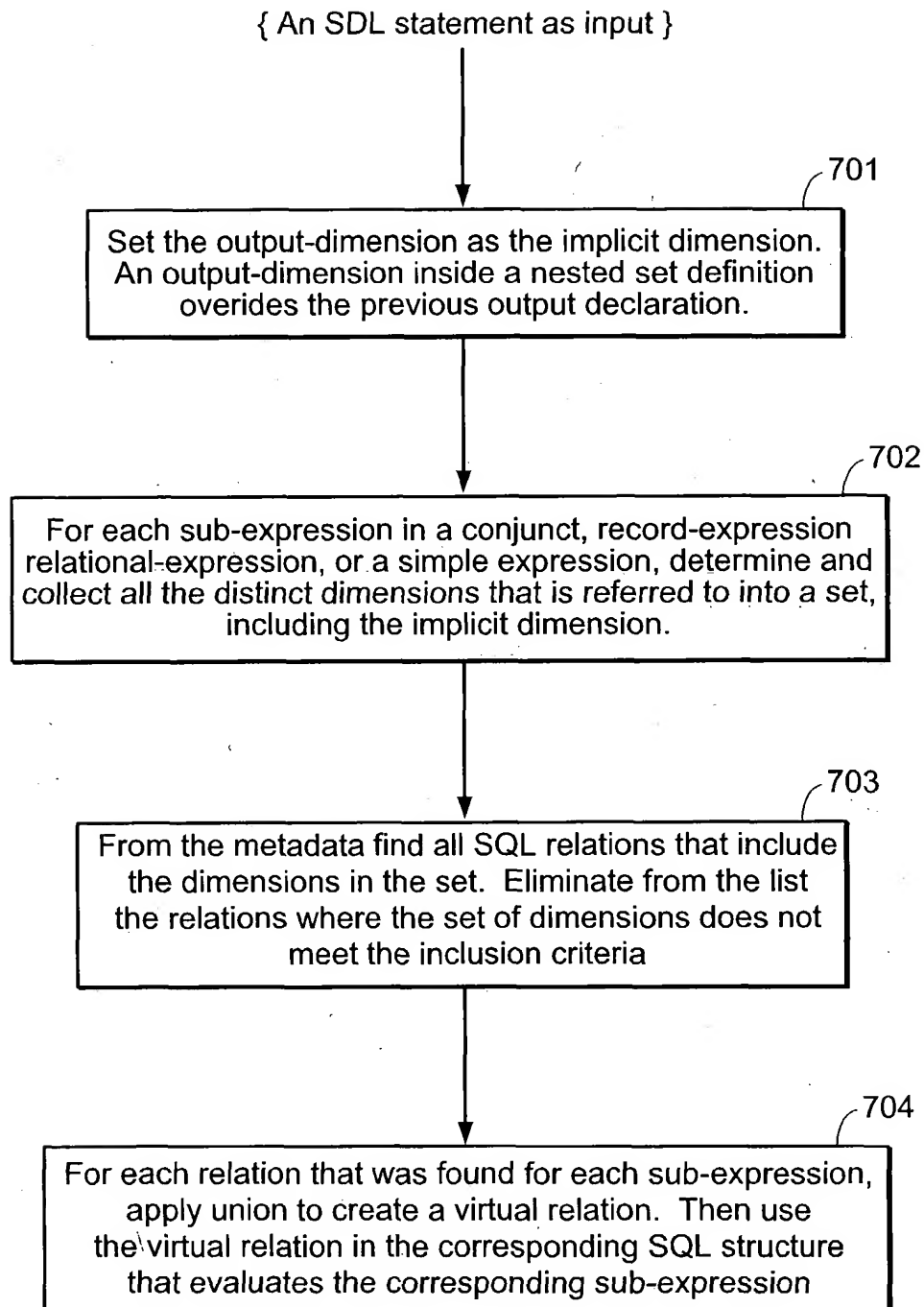


FIG. 7

8/35

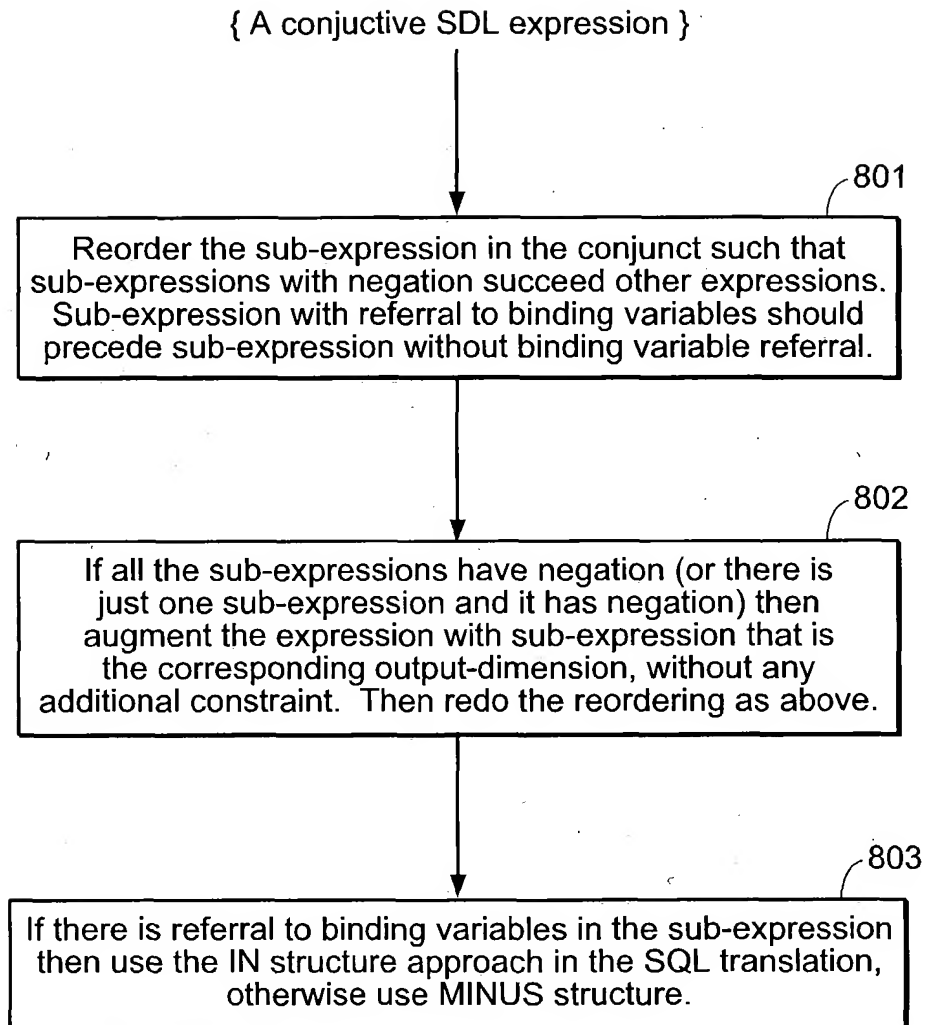


FIG. 8

9/35

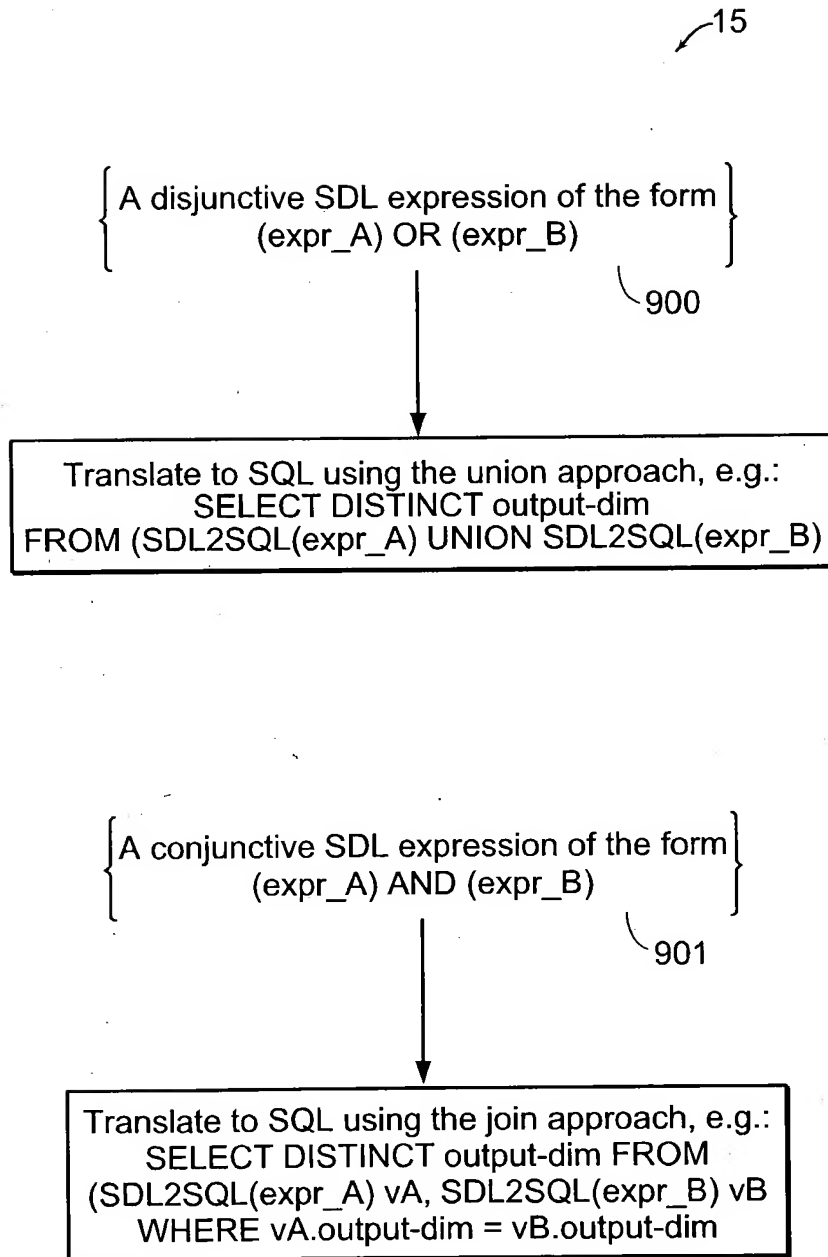


FIG. 9

10/35

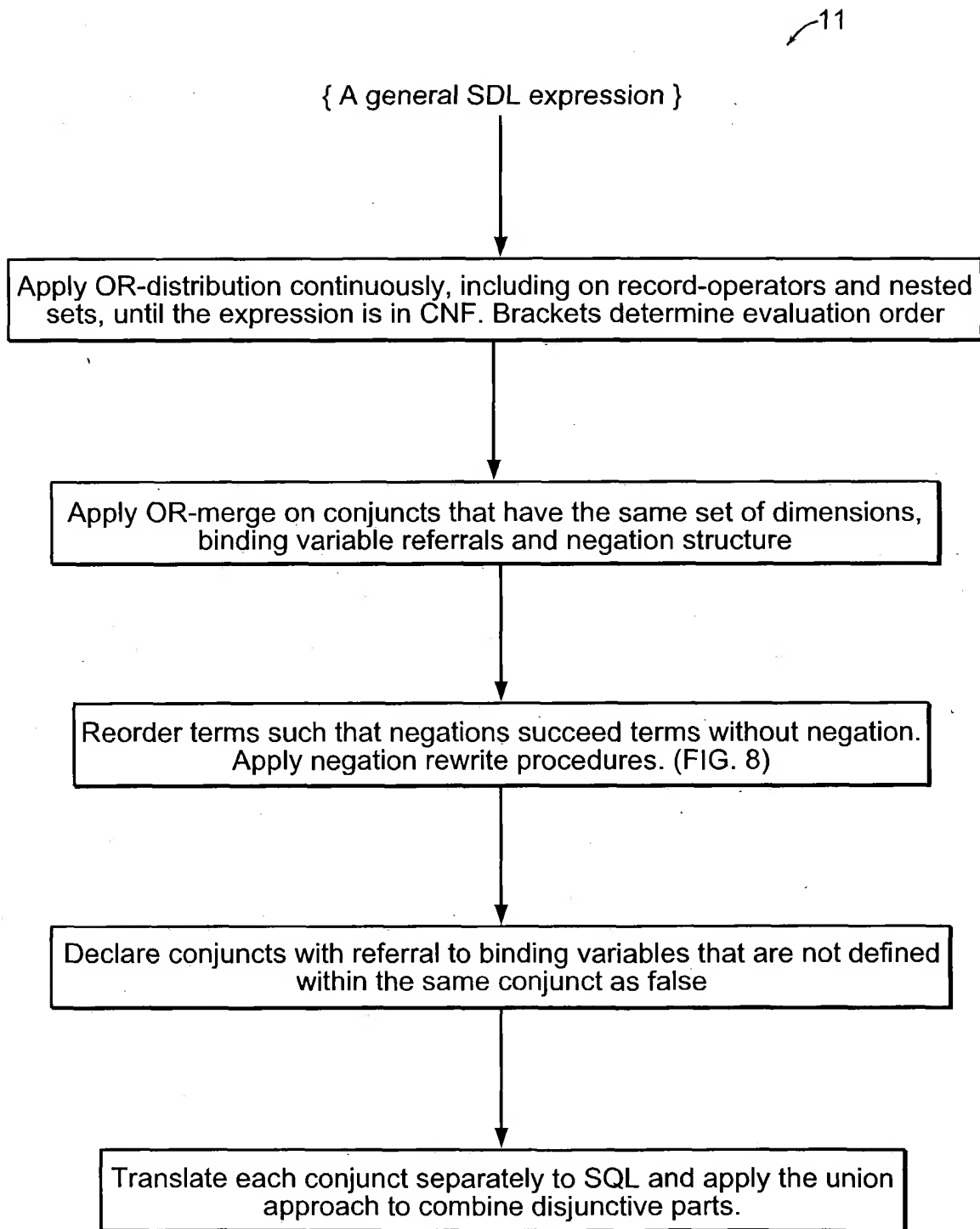


FIG. 10

11/35

13

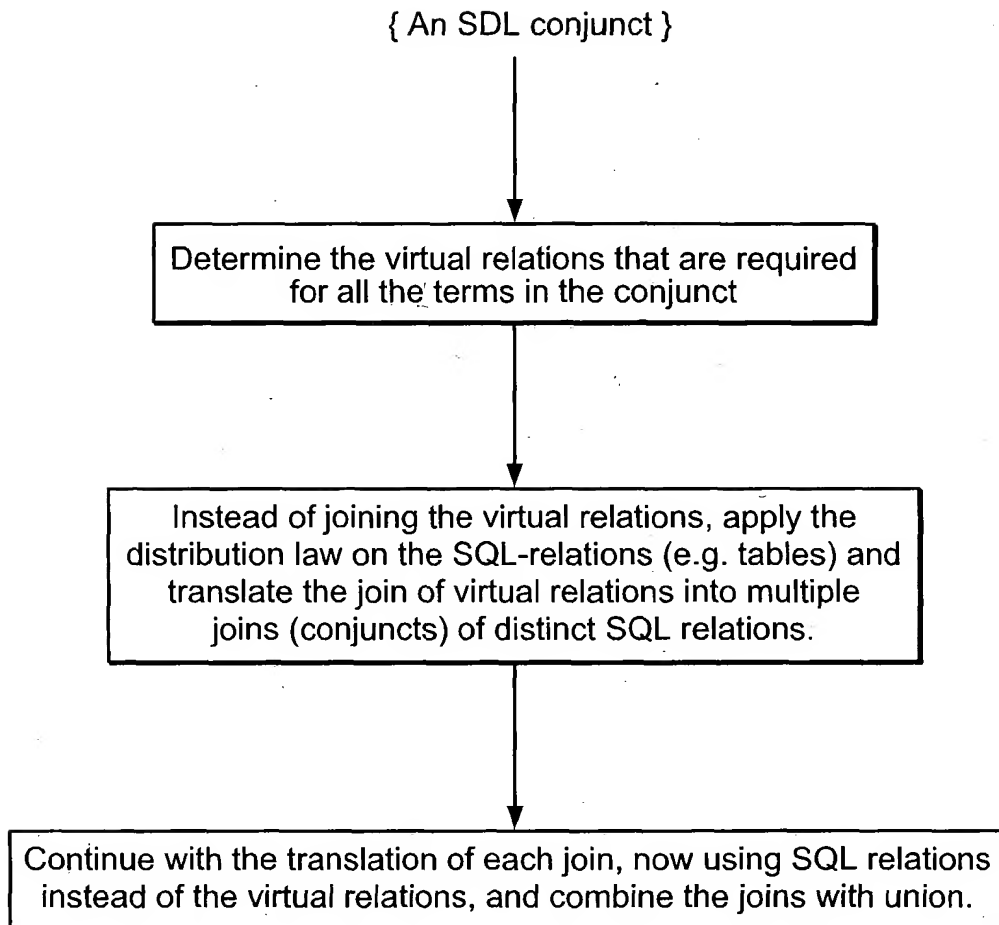


FIG. 11

12/35

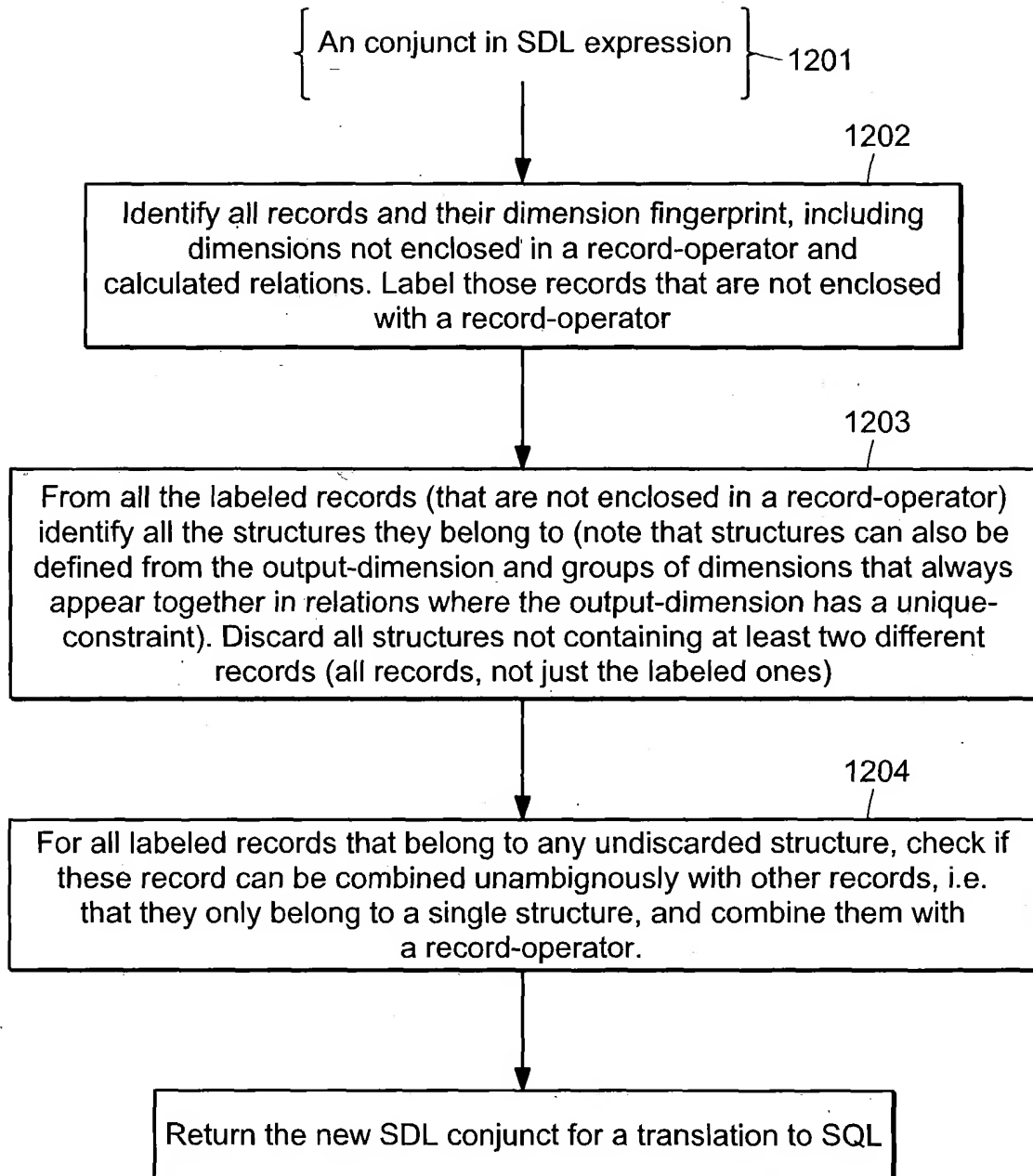


FIG. 12

13/35

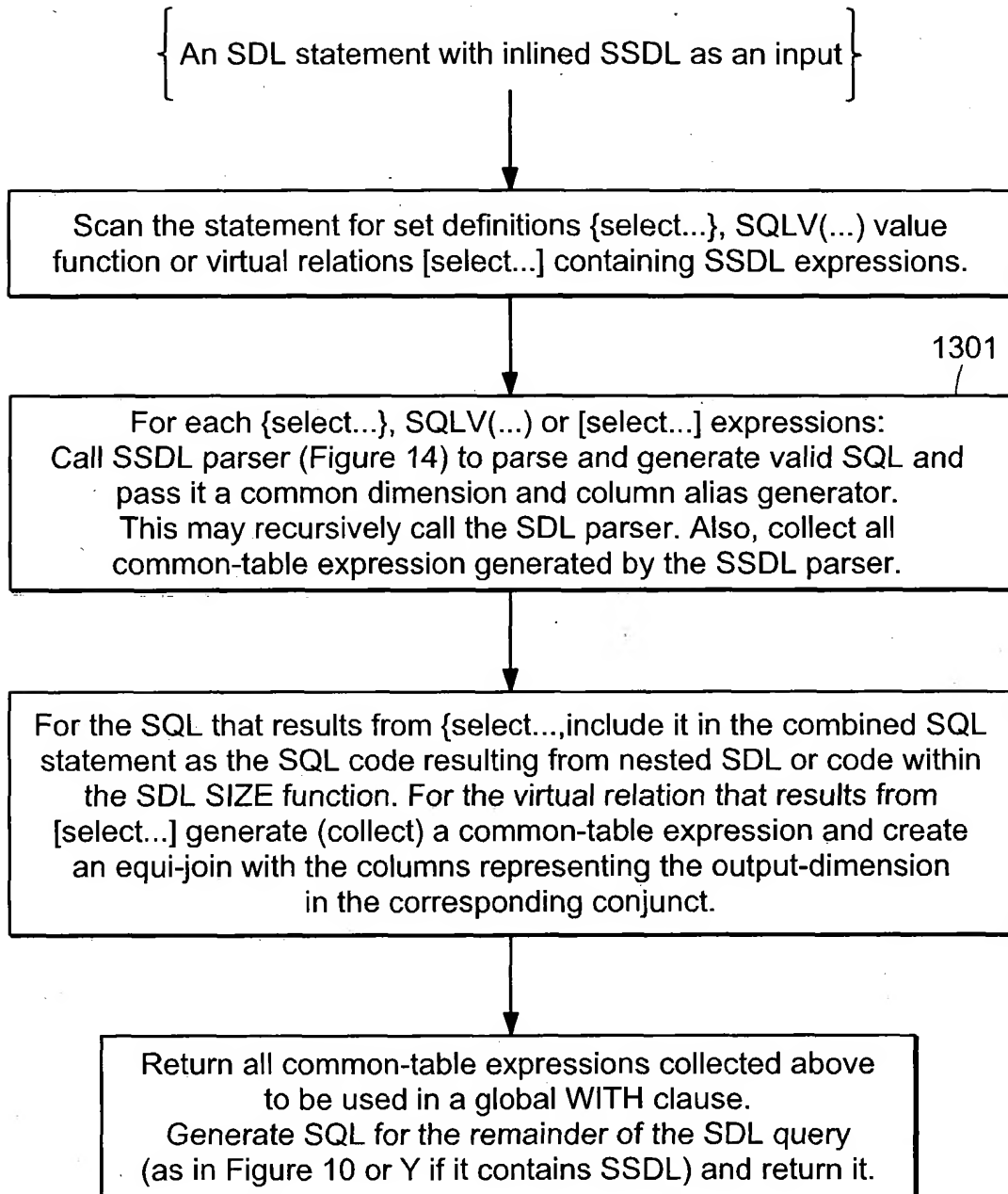


FIG. 13

14/35

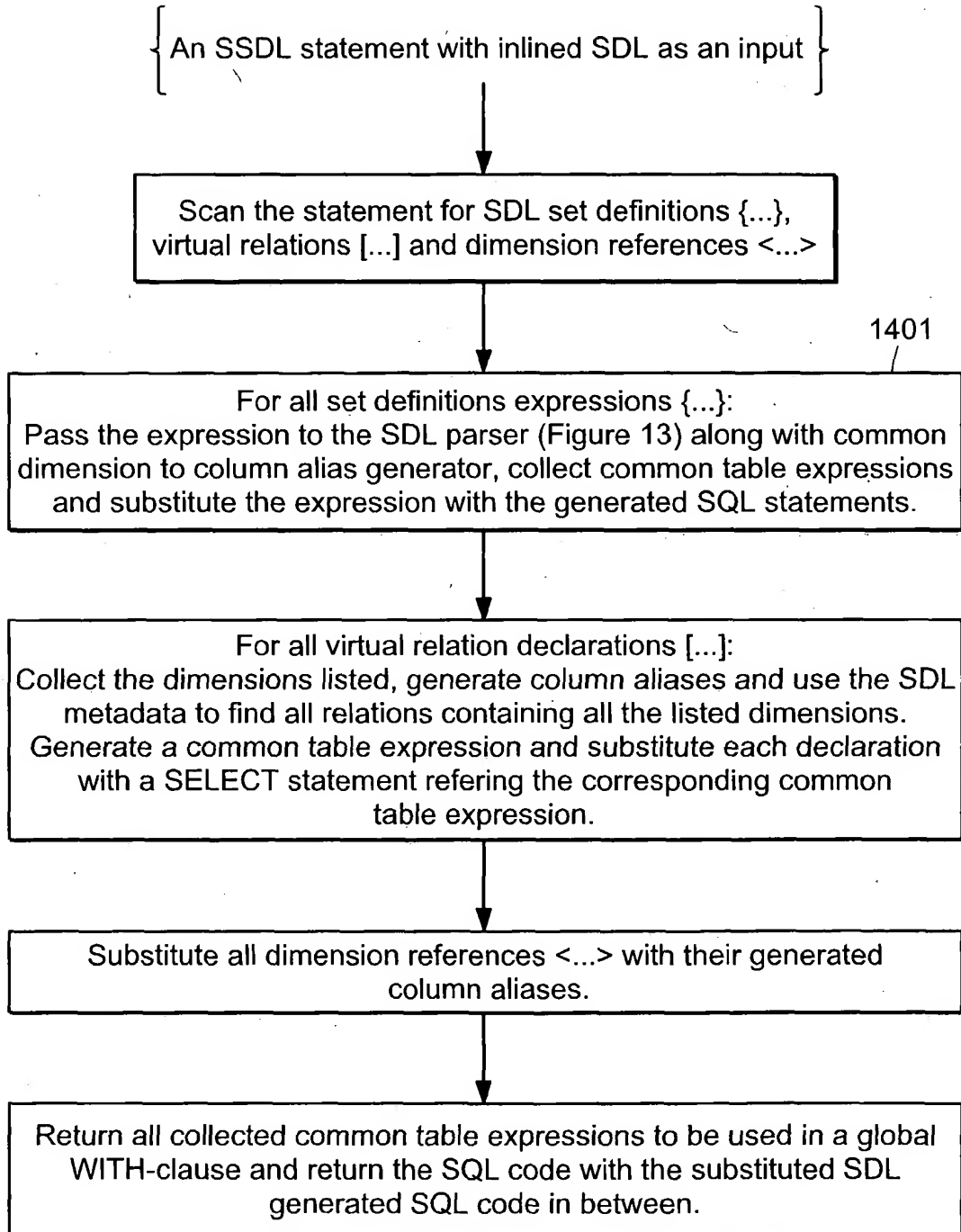


FIG. 14

15/35

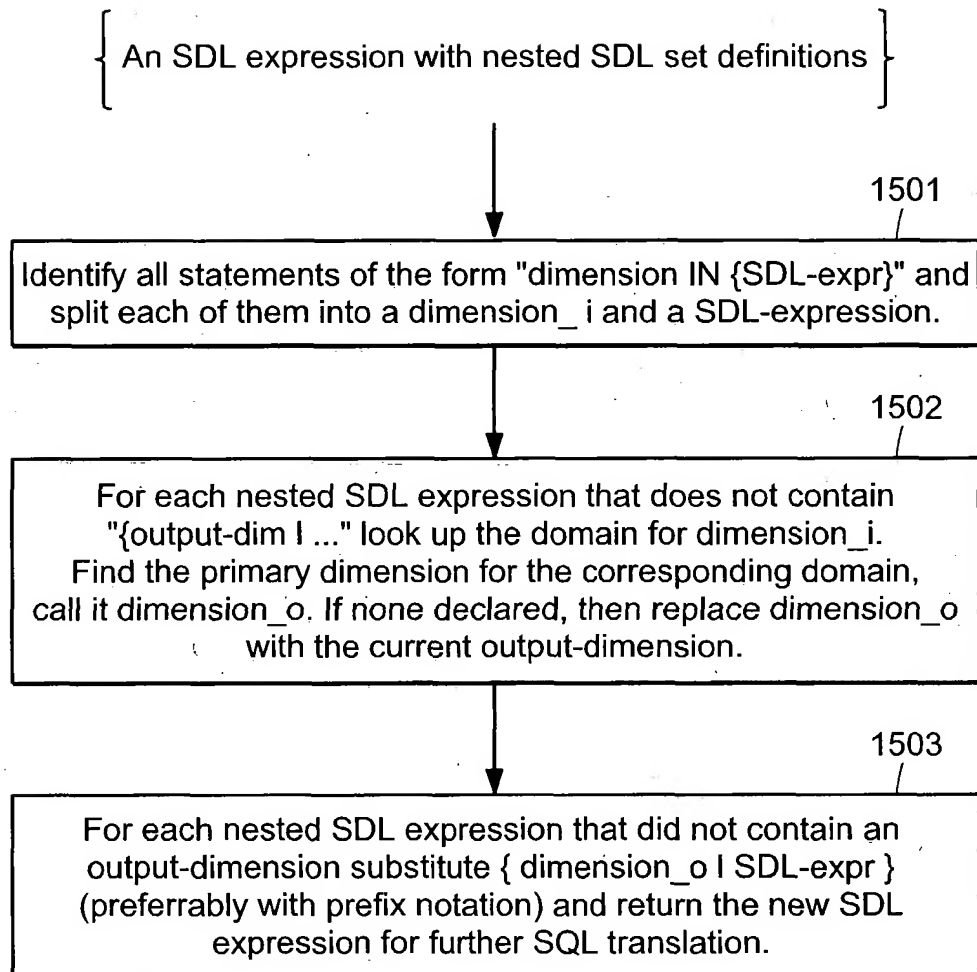


FIG. 15

16/35

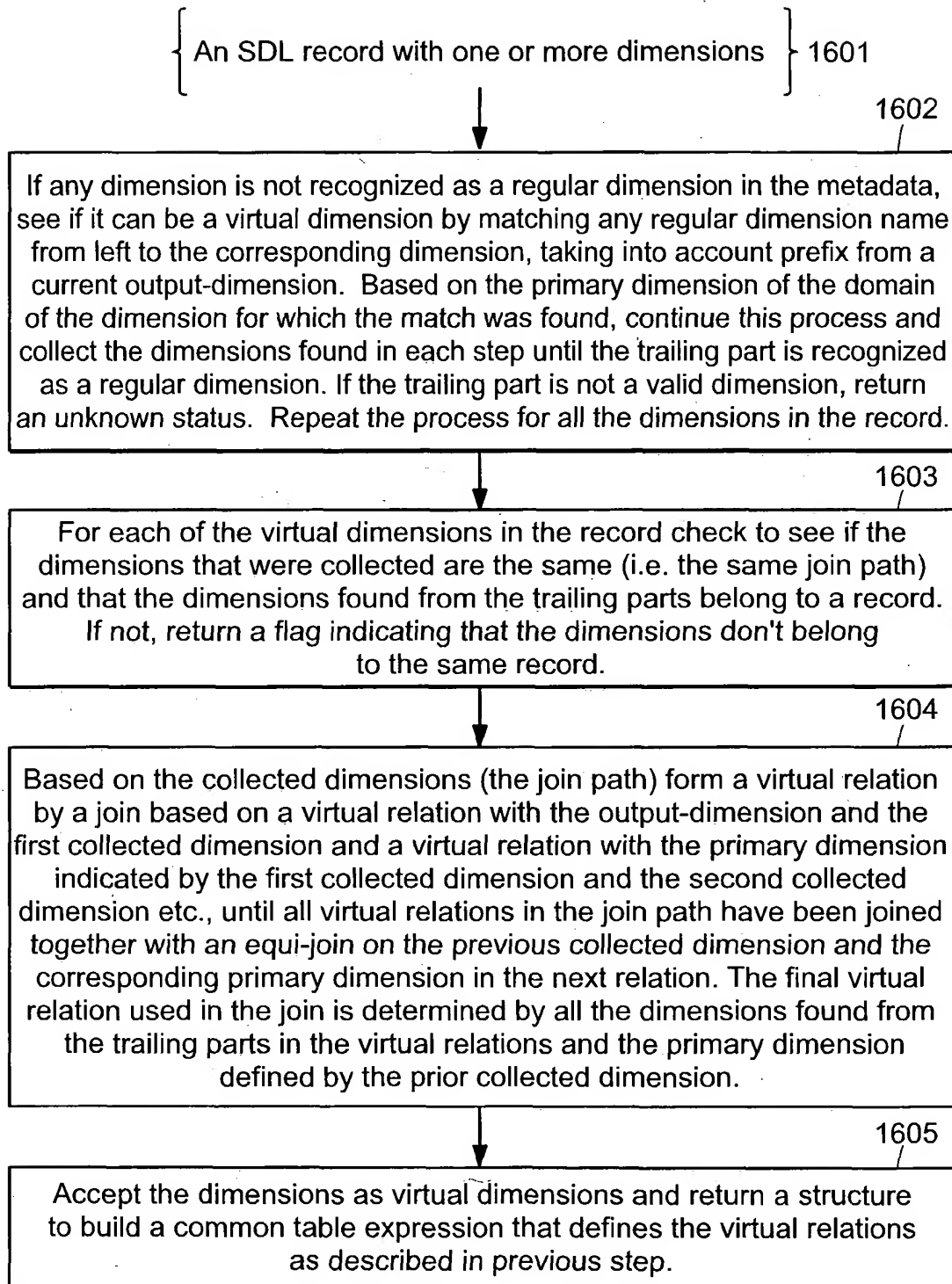


FIG. 16

17/35

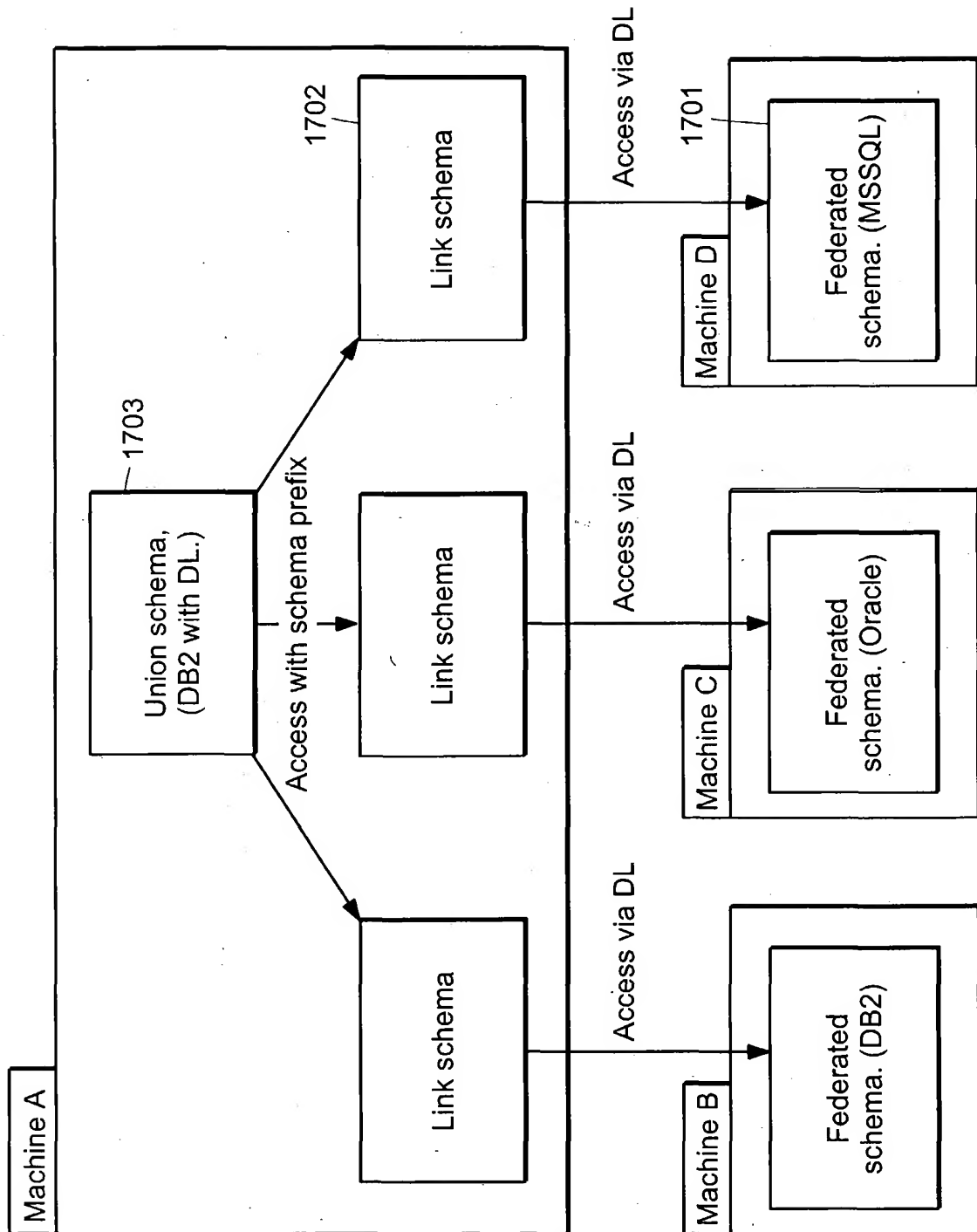


FIG. 17

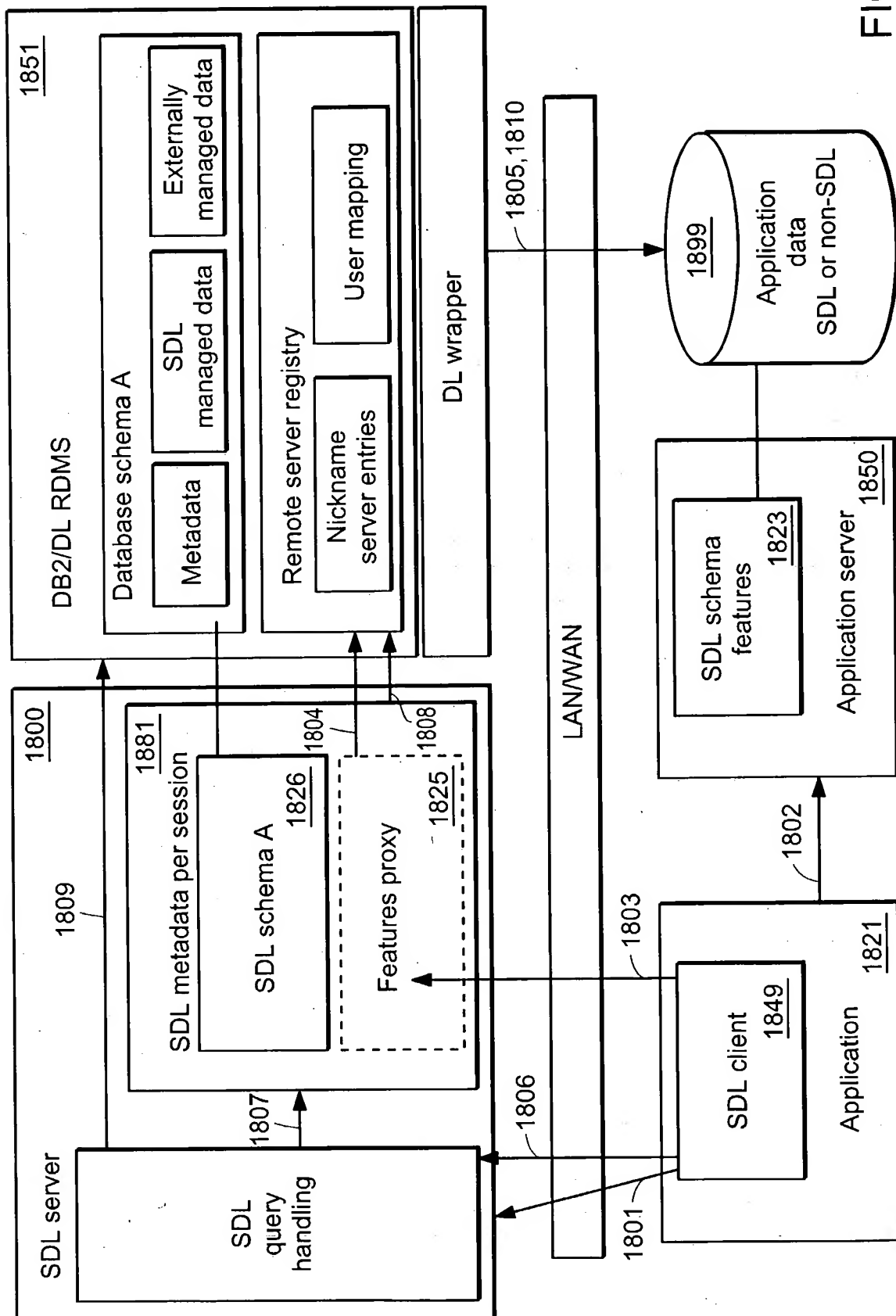


FIG. 18

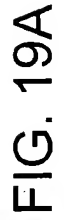


FIG. 19B

Query	Intersect
-------	-----------

Query Tool																												
<ul style="list-style-type: none"> <input type="checkbox"/> Projects <input checked="" type="checkbox"/> Gene Mining <input type="checkbox"/> Dimensions <input type="checkbox"/> Alternative gene IDs <input type="checkbox"/> Accession <input type="checkbox"/> Gene Symbol <input type="checkbox"/> Gene Symbol Alias <input type="checkbox"/> Expression <input type="checkbox"/> Gene ID <input type="checkbox"/> Gene Text Reports <input type="checkbox"/> Internal Systems dimensions <input type="checkbox"/> Linkage <input type="checkbox"/> Location <input type="checkbox"/> Mutations and Polymorphisms <input type="checkbox"/> Protein Structure and function <input type="checkbox"/> Enzyme Reactions <input type="checkbox"/> Functions <input type="checkbox"/> GeneOntology <input type="checkbox"/> Interaction <input type="checkbox"/> Pathways <input type="checkbox"/> PubGene <input type="checkbox"/> Medical Subject Headings <input type="checkbox"/> Medicine ID <input checked="" type="checkbox"/> Related Gene ID <input type="checkbox"/> Protein motifs <input type="checkbox"/> Sequence Similarity <input type="checkbox"/> Species Homology <input type="checkbox"/> Transcript related information <input type="checkbox"/> Resources 	<div style="border: 1px solid black; padding: 5px; margin-bottom: 10px;"> <p>PF.GO.TERM - :3673.8150.16265.8219 and PF.INTER.PUB.PGID</p> <div style="position: relative; height: 100px;"> 2100 </div> </div> <div style="border: 1px solid black; padding: 5px; margin-bottom: 10px;"> <p>Go Terms = :cell death and Related Gene ID</p> </div> <div style="margin-top: 20px;"> <div style="display: inline-block; border: 1px solid black; padding: 5px; margin-right: 10px;">Intersect</div> <div style="display: inline-block; border: 1px solid black; padding: 5px;">Query</div> </div>																											
<table border="1" style="width: 100%; border-collapse: collapse;"> <thead> <tr> <th>Name</th> <th>Expression</th> <th>Date</th> <th>Count</th> <th>Dimensions</th> </tr> </thead> <tbody> <tr> <td>Query 0</td> <td>ALT_IDS SYMBOL</td> <td>Wed Feb 06 17:00:20 GMT 2002</td> <td>14620</td> <td>GID</td> </tr> <tr> <td>Query 1</td> <td>ALT_IDS SYMBOL and NOT LOC.CHROM</td> <td>Wed Feb 06 17:01:00 GMT 2002</td> <td>47</td> <td>GID</td> </tr> <tr> <td>Query 2</td> <td>ALT_IDS SYMBOL_ALIAS</td> <td>Wed Feb 06 17:01:27 GMT 2002</td> <td>8128</td> <td>GID</td> </tr> <tr> <td>Query 3</td> <td>ALT_IDS SYMBOL and LOC.CHROW/</td> <td>Wed Feb 06 17:02:01 GMT 2002</td> <td>14573</td> <td>GID</td> </tr> </tbody> </table>				Name	Expression	Date	Count	Dimensions	Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID	Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID	Query 2	ALT_IDS SYMBOL_ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID	Query 3	ALT_IDS SYMBOL and LOC.CHROW/	Wed Feb 06 17:02:01 GMT 2002	14573	GID
Name	Expression	Date	Count	Dimensions																								
Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID																								
Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID																								
Query 2	ALT_IDS SYMBOL_ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID																								
Query 3	ALT_IDS SYMBOL and LOC.CHROW/	Wed Feb 06 17:02:01 GMT 2002	14573	GID																								
<div style="border: 1px solid black; padding: 5px;"> <p>Related Dimensions</p> <ul style="list-style-type: none"> <input type="checkbox"/> Gene ID <input type="checkbox"/> Medicine ID <input type="checkbox"/> Mesh String <input type="checkbox"/> Mesh Terms <input type="checkbox"/> Import Identifier </div>																												

FIG. 22A

FIG. 22B

FIG. 22

25/35

FIG. 22A

Query Tool

Projects
☐ Gene Mining
☐ Dimensions
☒ Alternative gene IDs
☐ Accession
☐ Gene Symbol
☐ Gene Symbol Alias
☐ Expression
☐ Gene ID
☐ Gene Text Reports
☐ Internal Systems dimensions
☐ Linkage
☐ Location
☐ Mutations and Polymorphisms
☐ Protein Structure and function
☐ Enzyme Reactions
☐ Functions
☐ Enzymes
☐ GPCR
☐ HMM
☐ Housekeeping
☐ Ion Channel
☐ GeneOntology
☐ Interaction
☐ Pathways
☐ PubGene
☐ Medical Subject Headings
☐ Medicine ID
☐ Related Gene ID
☐ Protein motifs
☐ Sequence Similarity
☐ Species Homology

Related Dimensions
☐ Gene ID
☐ Medicine ID
☐ Mesh String
☐ Mesh Terms
☐ Import identifier

SDL Editor External Venn Output Spec Dialog Builder Resources

PF.GO.TERM = :3673.8150.16265.8219 and PF.INTER.PUB.PGID .IN {PF.FUNC = :20.20}

2200

Go Terms = :cell death and Related Gene ID IN { Functions = :GPCP }

Intersect Query

Name	Expression	Date	Count	Dimensions
Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID
Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID
Query 2	ALT_IDS SYMBOL ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID
Query 3	ALT_IDS SYMBOL and LOC.CHROM/	Wed Feb 06 17:02:01 GMT 2002	14573	GID

2201

26/35

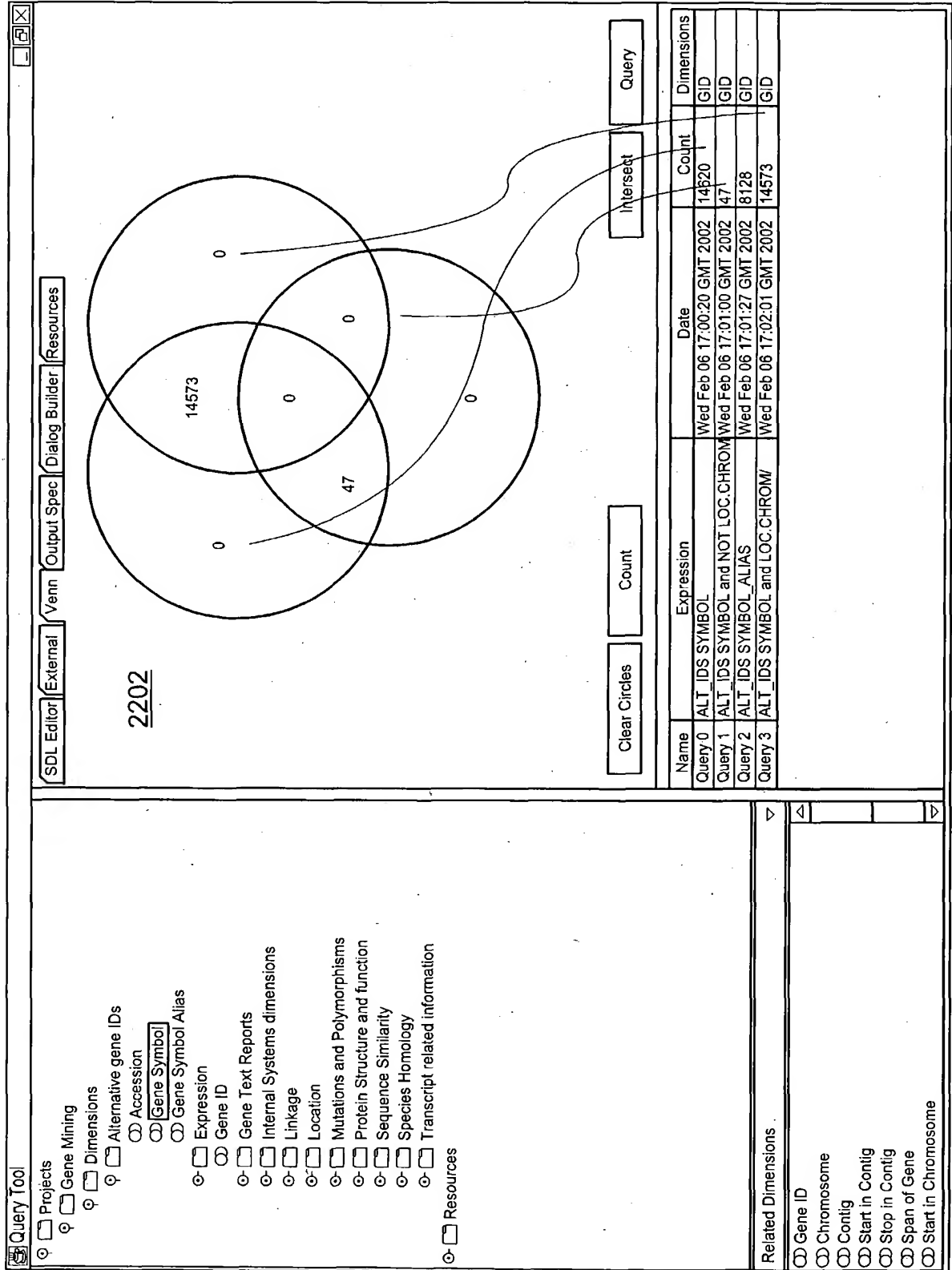


FIG. 22B

Docket No. 2345.2041-005

Docket No. 2345.2041-005

FIG. 23B

Query Tool

☐ Projects
☐ Gene Mining
☐ Dimensions
☐ Alternative gene IDs
☐ Accession
☐ Gene Symbol
☐ Gene Symbol Alias
☐ Expression
☐ Gene ID
☐ Gene Text Reports
☐ Gene Description
☐ Locus Link Text Summary
☐ Internal System dimensions
☐ Linkage
☐ Disease
☐ Lod Score
☐ Phenotype
☐ Location
☐ Mutations and Polymorphism
☐ Protein Structure and function
☐ Enzyme Reactions
☐ Functions
☐ Enzymes
☐ GPCR
☐ HMM
☐ Housekeeping
☐ Ion Channel
☐ Gene Ontology
☐ Interaction
☐ Pathways
☐ PubGene
☐ Medical Subject Headings

☐ Related Dimensions
☐ Gene ID
☐ Lod Score
☐ Phenotype
☐ Import Identifier

SDL Editor | **External** | **Venn** | **Output Spec** | **Dialog Builder** | **Resources**

Output Mode
☒ Outer Join **2402** ☒ Strict relation **2403**

Output Dimensions

@	Dimension	SDLQueryOutputFunction
0	GID	Group by
1	TEXT.DESC	Group by
2	LINK.DIS	Count distinct

2400

Calculated Columns

Description	Pattern

Name	Expression	Date	Count	Dimensions
Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID
Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID
Query 2	ALT_IDS SYMBOL_ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID
Query 3	[ALT_IDS SYMBOL and LOC.CHROM]	Wed Feb 06 17:02:01 GMT 2002	14573	GID
Query 4	PF.GO.TERM= 3673.8150.16265.8219 ...	Wed Feb 06 17:19:18 GMT 2002	58	GID

FIG. 24A

FIG. 24B

FIG. 24

29/35

FIG. 24A

①

30/35

①

SDL query [58]			COUNT(DISTINCT LINK.DIS	
(GID %1)			2401	
GID	TEXT.DE8C	COUNT(DISTINCT LINK.DIS		
Hs.101174	microtubule-associated protein tau	9		
Hs.105658	DNA fragmentation factor, 45 kD, alpha polyp...	0		
Hs.11103	transforming growth factor, beta 1	4		
Hs.11171	APG5 (autophagy5, 6, cerevisiae)-like	0		
Hs.12409	somatostatin	8		
Hs.128087	coagulation factor II (thrombin receptor	7		
Hs.1313	tumor necrosis factor (ligand) superfamily m...	8		
Hs.133089		0		
Hs.142255	islet amyloid polypeptide	6		
Hs.146355	v-abl Aberson murine leukemia viral oncogen...	8		
Hs.151988	mitogen-activated protein kinase kinase kina...	10		
Hs.159494	Bruton agammaglobulinemia tyrosine kinase	0		
Hs.1613	adenosine A2a receptor	6		
Hs.170917	prostaglandin E receptor 3 (subtype EP3)	10		
Hs.1722	interleukin 1, alpha	0		
Hs.1724	interleukin 2 receptor, alpha	0		
Hs.172894	BH3 interacting domain death agonist	5		
Hs.176090	PRKC, apoptosis, WT1, regulator	7		
Hs.177486	amyloid beta (A4) precursor protein (protease...	6		
Hs.180338	tumor necrosis factor receptor superfamily, m...	9		
Hs.1817	myeloperoxidase	8		
Hs.1846	tumor protein p53 (Li-Fraumeni syndrome)	8		
Hs.2007	tumor necrosis factor (ligand) superfamily, m...	9		
Hs.20313	protein tyrosine kinase 2 beta	6		
Hs.21486	signal transducer and activator of transcrip...	8		
Hs.225995	somatostatin receptor 3	6		
Hs.239176	insulin-like growth factor 1 receptor	4		
Hs.239489	TIA1 cytotoxic granule-associated RNA-bindin...	8		
58 rows (Total: 4426 msec)			Abort	

FIG. 24B

FIG. 25A

FIG. 25B

FIG. 25

31/35

FIG. 25A

Query Tool

- ☐ Projects
- ☐ Gene Mining
- ☐ Dimensions
 - ☐ Alternative gene IDs
 - ☐ Accession
 - ☐ Gene Symbol
 - ☐ Gene Symbol Alias
 - ☐ Expression
 - ☐ Gene ID
 - ☐ Gene Text Reports
 - ☐ Gene Description
 - ☐ Locus Link Text Summary
 - ☐ Internal Systems dimensions
 - ☐ Linkage
 - ☐ Disease
 - ☐ Lod Score
 - ☐ Phenotype
 - ☐ Location
 - ☐ Mutations and Polymorphisms
 - ☐ Protein Structure and function
 - ☐ Enzyme Reactions
 - ☐ Functions
 - ☐ Enzymes
 - ☐ GPCR
 - ☐ HMM
 - ☐ Housekeeping
 - ☐ Ion Channel
 - ☐ GeneOntology
 - ☐ Interaction
 - ☐ Pathways
 - ☐ PubGene
 - ☐ Medical Subject Headings

2500

Test dialog
Disease association
Lodscore calculated with
Allegra

2502

☐ LINK.DIS = Disease

☐ LINK.LODS = Lod.Score

2501

☐ AND

SDI Editor **External** **Venn** **Output Spec** **Dialog Builder** **Resources**

Name	Expression	Date	Count	Dimensions
Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID
Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID
Query 2	ALT_IDS SYMBOL ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID
Query 3	ALT_IDS SYMBOL and LOC.CHROM/	Wed Feb 06 17:02:01 GMT 2002	14573	GID
Query 4	PF GO TERM = 3673,8150,16265,8219,	Wed Feb 06 17:19:18 GMT 2002	58	GID

Related Dimensions

- ☐ Gene ID
- ☐ Disease
- ☐ Phenotype
- ☐ Import identifier

32/35

FIG. 25B

Query Tool

- Projects
 - Gene Mining
 - Dimensions
 - Alternative gene IDs
 - Accession
 - Gene Symbol
 - Gene Symbol Alias
 - Expression
 - Gene ID
 - Gene Text Reports
 - Gene Description
 - Locus Link Text Summary
 - Internal Systems dimensions
 - Linkage
 - Disease
 - Lod Score**
 - Phenotype
 - Location
 - Mutations and Polymorphisms
 - Protein Structure and function
 - Enzyme Reactions
 - Functions
 - Enzymes
 - GPCR
 - HMM
 - Housekeeping
 - Ion Channel
 - GeneOntology
 - Interaction
 - Pathways
 - PubGene
 - Medical Subject Headings

Gene Mining

2504

2503

Test dialog
Lod scores calculated with Allegro

Disease = AND

Lod Score = AND

Name	Expression	Date	Count	Dimensions
Query 0	ALT_IDS SYMBOL	Wed Feb 06 17:00:20 GMT 2002	14620	GID
Query 1	ALT_IDS SYMBOL and NOT LOC.CHROM	Wed Feb 06 17:01:00 GMT 2002	47	GID
Query 2	ALT_IDS SYMBOL ALIAS	Wed Feb 06 17:01:27 GMT 2002	8128	GID
Query 3	ALT_IDS SYMBOL and LOC.CHROM/	Wed Feb 06 17:02:01 GMT 2002	14573	GID
Query 4	PE.GO.TERM = 3673.8150.16265.8219	Wed Feb 06 17:19:18 GMT 2002	58	GID
Query 5	LINK.DIS + Alzheimers AND LINK.LOD...	Wed Feb 06 17:30:51 GMT 2002	126	GID

Related Dimensions

- Gene ID
- Disease
- Phenotype
- Import identifier

Data Management Tool			
<input type="checkbox"/> Projects	<input type="checkbox"/> Manage imports	<input type="checkbox"/> Edit import	<input type="checkbox"/> Link Table XML
<input checked="" type="checkbox"/> Dimensions			Hierarchy Import URL Template
<input checked="" type="checkbox"/> Diagnose	Name	Description	URL
<input type="checkbox"/> Internal System dimensions	Diagnosis Web Data.. This Web provides details on ICD9 Codes http://www.diagweb.org/cgi... DIAGN		
<input checked="" type="checkbox"/> Person identifier			
<input checked="" type="checkbox"/> Person identifier			
<input checked="" type="checkbox"/> Person identifier B			
<input type="checkbox"/> Project Codes			
<input type="checkbox"/> Measurement			
<input checked="" type="checkbox"/> Bloodpressure High			
<input checked="" type="checkbox"/> Bloodpressure Low			
<input type="checkbox"/> Nice Strings			
<input type="checkbox"/> Project Codes 2			
<input type="checkbox"/> Domains			
<input checked="" type="checkbox"/> Amount			
<input type="checkbox"/> ICD-10 International classification			
<input checked="" type="checkbox"/> Integer			
<input checked="" type="checkbox"/> Long String			
<input checked="" type="checkbox"/> Person identifier			
<input checked="" type="checkbox"/> Person identifier 2			
<input checked="" type="checkbox"/> Sample id			
<input checked="" type="checkbox"/> Small integer			
<input checked="" type="checkbox"/> String			

Diagnosis Web Data Bank			
Name	This Web provides details on ICD9 Codes — 2603		
Description	http://www.diagweb.org/cgi-bin/find_icd? code=%DIAGN% — 2602		
Template	DIAGN — 2601		

Related Dimensions			
<input checked="" type="checkbox"/>	Bloodpressure High		
<input checked="" type="checkbox"/>	Bloodpressure Low		
<input checked="" type="checkbox"/>	Changed Second String		
<input checked="" type="checkbox"/>	First String		
<input checked="" type="checkbox"/>	Import identifier		
<input checked="" type="checkbox"/>	Person identifier		

34/35

Diagnose

PID	DIAGN	CALC
PID0	A.04.0	abc PID0 def
PID1	A.04.1	abc PID1 def
PID2	A.04.2	abc PID2 def
PID3	A.04.3	abc PID3 def
PID4	A.04.4	abc PID4 def
PID5	A.04.5	abc PID5 def
PID6	A	abc PID6 def
PID7	A	abc PID7 def
PID8	A	abc PID8 def
PID9	A	abc PID9 def

Search and Select

Apply Selection

Reset Selection

Invert Selection

URL Templates

2604

This Web provides details on ICD9 Codes.

Network Error - Microsoft Internet Explorer provided by deCODE Genetics, Inc

File

Edit

View

Favorites

Tools

Help

Back

Forward

Stop

Home

Search

Favorites

History

Address

http://www.diagweb.org/cdi-bin/find_icd? code=A.04.6

Go

Links

10 rows (Total: 1142 msec)

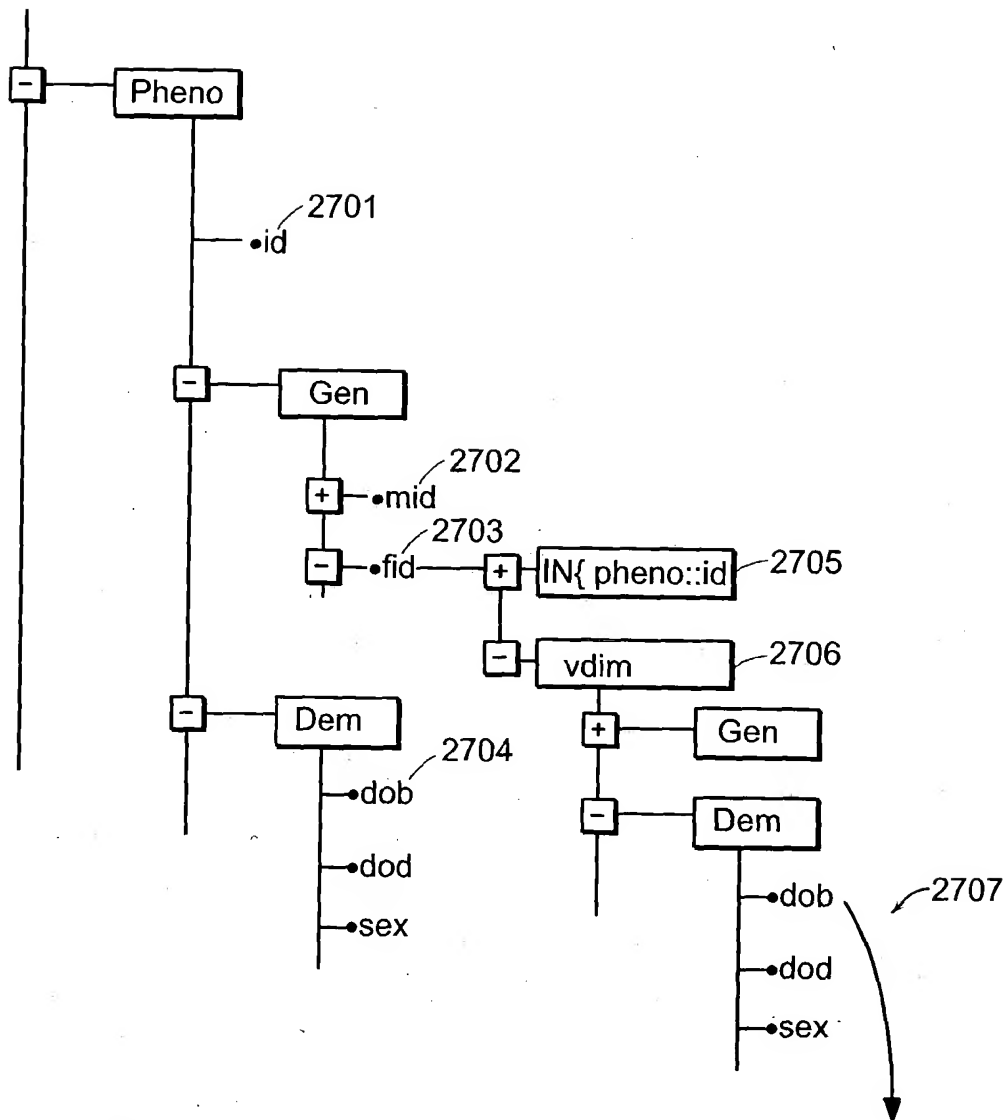
2605

Abort

Close

FIG. 26B

35/35



SDL editor

{ pheno :: id | gen. fid.dem.dob > 2000 } 2708

{ pheno :: id | gen. fid IN { dem.dob > 2000 } } 2709

FIG. 27